MTMR2 gene

myotubularin related protein 2

Normal Function

The *MTMR2* gene provides instructions for making an enzyme called myotubularin related protein 2. This enzyme is a phosphatase, which means it helps remove a phosphate group (a cluster of one phosphorus atom and three oxygen atoms) from other substances. Removal of a phosphate group modifies the activity of these substances.

The MTMR2 protein modifies substances that act as chemical messengers. These messengers relay signals from receptors on the cell surface to specific compartments inside the cell, through a process called signal transduction. Signal transduction helps cells respond to their environment, for example, by dividing or maturing to take on specialized functions. The MTMR2 protein modifies chemical messengers that help regulate processes such as the transport of fats (lipids) and proteins within the cell.

Health Conditions Related to Genetic Changes

Charcot-Marie-Tooth disease

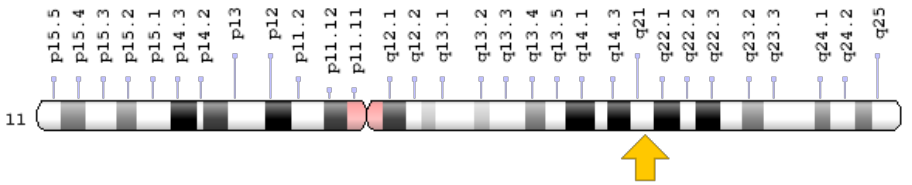
Researchers have identified at least 10 *MTMR2* gene mutations that cause a form of Charcot-Marie-Tooth disease known as type 4B1. Some of these mutations create a premature stop signal in the instructions for making the MTMR2 protein, which results in an abnormally short protein. Other mutations alter the protein's structure by changing one of the building blocks (amino acids) used to make the protein. All of these mutations probably impair the protein's ability to modify chemical messengers, which could disrupt the transport of lipids and proteins.

It is unclear how *MTMR2* gene mutations lead to the characteristic features of type 4B1 Charcot-Marie-Tooth disease. Scientists suggest that specialized cells in the nervous system, called Schwann cells, are particularly vulnerable when the MTMR2 protein is impaired. Schwann cells produce myelin, a protective substance that covers nerves and promotes the rapid transmission of nerve impulses. Schwann cells use large amounts of lipids and proteins to make myelin, relying on efficient transport of these substances within cells. Disrupted transport could affect myelin production, altering the transmission of nerve impulses. A disturbance in nerve impulse transmission is a sign of type 4B1 Charcot-Marie-Tooth disease.

Chromosomal Location

Cytogenetic Location: 11q21, which is the long (q) arm of chromosome 11 at position 21

Molecular Location: base pairs 95,832,880 to 95,924,207 on chromosome 11 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- CMT4B
- KIAA1073
- MTMR2_HUMAN

Additional Information & Resources

Educational Resources

- Biochemistry (fifth edition, 2002): Signal-Transduction Pathways
  https://www.ncbi.nlm.nih.gov/books/NBK21205/

GeneReviews

- Charcot-Marie-Tooth Neuropathy Type 4
  https://www.ncbi.nlm.nih.gov/books/NBK1468

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28MTMR2%5BTIAB%5D%29+OR+%28myotubularin+related+protein+2%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2520+days%22%5Bdp%5D

OMIM

- MYOTUBULARIN-RELATED PROTEIN 2
  http://omim.org/entry/603557

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_MTMR2.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=MTMR2%5Bgene%5D
- HGNC Gene Family: Myotubularins
  http://www.genenames.org/cgi-bin/genefamilies/set/903
- HGNC Gene Family: Phosphoinositide phosphatases
  http://www.genenames.org/cgi-bin/genefamilies/set/1079
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7450
- Inherited Peripheral Neuropathies Mutation Database
  http://www.molgen.ua.ac.be/CMTMutations/Mutations/Mutations.cfm?Context=6
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/8898
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